VDH VIRGINIA DEPARTMENT OF HEALTH

Division of Child and Family Health- Newborn Screening Follow-Up Program

# Biotinidase deficiency (BIOT)

*Parent Fact Sheet*

A newborn screening test is a **screen** and not diagnostic testing. An "abnormal" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

**Disorder Indicated:** Biotinidase deficiency (BIOT) is an inherited condition in which the body is unable to reuse and recycle the vitamin biotin. Because the body needs free biotin to break down fats, proteins, and carbohydrates effectively, individuals with BIOT are less able to process important nutrients.

Children with BIOT who are identified through newborn screening and begin treatment immediately usually remain healthy with normal development. Some children with mild forms may not need any treatment.

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| Incidence | 1 in every 60,000 newborns. |
| Signs and Symptoms<br><br>*Please note: these findings may not be present in young infants or in milder forms of the disease* | When a child has Biotinidase deficiency, you may see symptoms including:<br>• Seizures<br>• Weak muscle tone (hypotonia)<br>• Trouble breathing<br>• Skin rash<br>• Hair loss<br>• Trouble balancing<br>• A fungal infection called candidiasis<br><br>*Many of these signs can be triggered by illnesses or infections.* |
| Next Steps ***may*** include: | **Follow up with your child's pediatric provider**<br>Clinical assessment<br>Laboratory Testing |
| Treatment ***(if indicated)*** | Change in diet (addition of Biotin supplements) |
| Additional Resources | VDH Newborn Screening *http://vdhlivewell.com/newbornscreening*<br>Baby's First Test *www.babysfirsttest.org*<br>Genetics Home Reference *https://ghr.nlm.nih.gov/*<br>Biotinidase Deficiency Support Group<br>*www.biotinidasedeficiency.20m.com/* |

*Educational content adapted from www.babysfirsttest.org*

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